

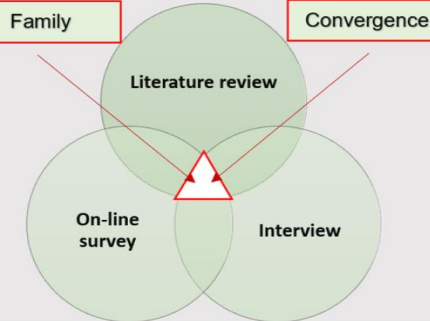
PhD Study Aim

To explore the relationship between Quality of Life and Social Support for adults living with rare genetic skin conditions.

Participants

Thirty-one adults with diagnosed rare genetic skin and connective tissues conditions including Ehlers-Danlos Syndrome (EDS) and Epidermolysis Bullosa (EB).

Triangulation methodology

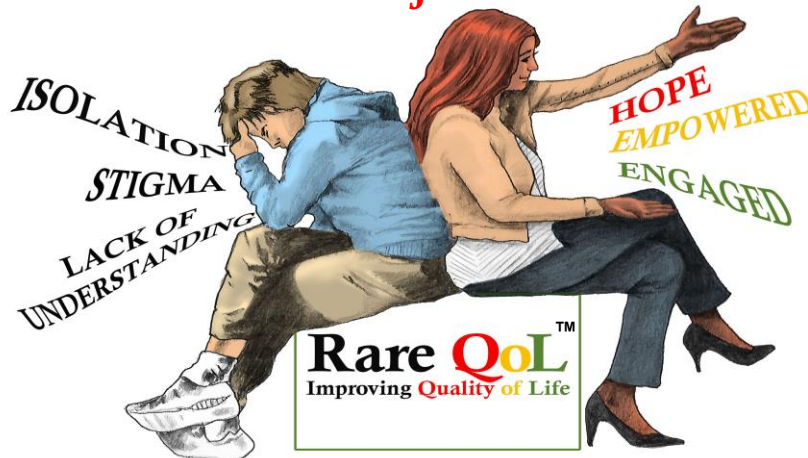


Impact of Diagnosis

“Once you get a diagnosis, that is more or less where the support stops”.

“It affected me massively. I just went completely off the rails....I just completely shut down”.

Stories are not just for bed time



Quality of Life Measures

QoL measures were compared. All included physical, psychological and social dimensions. Most rare disease research is focused on the physical dimensions.

QUALITY OF LIFE MEASURES	DIMENSIONS					
	Physical	Psychological	Social	Environment	Independence	Spirituality
Children's Dermatology Life Quality Index	Yes	Yes	Yes	No	No	No
Dermatology Life Quality Index	Yes	Yes	Yes	No	No	No
Euro Quality of Life 5 Dimension Questionnaire	Yes	Yes	Yes	No	No	No
Family Dermatology Life Quality Index	Yes	Yes	Yes	No	No	No
Quality of Life in Epidermolysis Bullosa	Yes	Yes	Yes	No	No	No
Short Form 36	Yes	Yes	Yes	No	No	No
SKINDEX	Yes	Yes	Yes	No	No	No
World Health Organisation Quality of Life Questionnaire	Yes	Yes	Yes	Yes	Yes	Yes

Outcomes

- Some participants reported that they felt stigmatised, a sense of social isolation and experienced difficulties in managing the emotional impact of their condition.
- Analysis indicated a positive correlation between QoL and Social Support.
- The majority of the emotional support was provided by the family.

Conclusion

Most rare disease research is focused on the physical aspects of conditions. Current world issues has taught us that adopting a holistic bio-psycho-social approach has to be a priority to effectively support the Rare Disease Community.

PhD Study Supervisors

Professor Elizabeth Mason -Whitehead
Associate Professor Dr Andrew Mitchell

Sondra Butterworth

Final stage PhD Student
MBPsS., MSc. Psychology
Board member Genetic Alliance UK
Community Psychology Specialist
sondra.butterworth@rareqol.org.uk